State of California – Health and Human Services Agency
Department of Health Services

California Department of Health Services

SANDRA SHEWRY
Director

ARNOLD SCHWARZENEGGER
Governor

October 12, 2004

Dear Health Care Provider:

Legislation authored by Senator Alpert and signed by the Governor has authorized the expansion of the newborn screening program to include disorders detectable by Tandem Mass Spectrometry (MS/MS), i.e., fatty acid oxidation disorders, organic acid disorders and amino acid disorders. The 18 month MS/MS pilot project conducted by the Department found 1 in every 6,500 newborns tested had one of these disorders. Enclosed is a list of disorders that can be detected, based on the analytes that will be measured. The law also includes screening for congenital adrenal hyperplasia. These additional disorders will not be screened for or included in the reports to physicians until the state program is operational. We will send another letter announcing the starting date and providing more details about the expanded screening but the legislation requires statewide screening by August 1, 2005.

In the interim, we continue to urge you to provide information to all pregnant women about the disorders not included in the state program and how to obtain supplemental testing. We provide a booklet entitled *Important Information for Parents about The Newborn Screening Test*, which prenatal care providers and maternity hospitals are required by law to distribute. In addition to containing information on the current mandatory newborn screening test, the most recent version (June 2004) includes information on supplemental screening and a list of the private laboratories that offer the testing. Additional copies of this booklet can be obtained free of charge by calling the newborn screening educational materials order line at (510) 412-1542. Information on optional supplemental screening is also available on our website (www.dhs.ca.gov/gdb then click on newborn screening). In addition, for a nominal fee, you can purchase copies of the pamphlet *A Simple Test Could Save Your Baby's Life* from Save Babies Through Screening Foundation. This pamphlet includes information for expectant parents on how to obtain supplemental screening. A sample of the pamphlet, is enclosed. Save Babies Through Screening Foundation can be contacted at: 1-888-454-3383, or email@savebabies.org. The pamphlet, in English or Spanish, can also be downloaded from their website at www.savebabies.org.

The supplemental tests are not a covered Medi-Cal benefit and patients should be informed that their insurance might not cover the laboratory charge. It will also require the collection of two specimens, one using the State collection form which is submitted to a State contract laboratory for the mandatory testing and the second on a collection

kit form supplied by the private laboratory which is sent to them for testing. Be careful not to mix up or misdirect these forms.

If patients or hospitals choose to use one of these private laboratories and have a positive result, expert consultation in deciding on diagnostic testing and interpreting the results to the parents is available from the state approved metabolic centers. A list of contacts is enclosed.

The private laboratories will not be reporting supplemental screening results to the Newborn Screening Program, and the Area Service Center Newborn Screening Coordinators will not be involved in tracking of follow-up for newborns with inadequate or positive results on supplemental screening done through the private laboratories.

We appreciate that this separate offering of supplemental testing is an additional burden, but the inconvenience will be temporary until the statewide program starts. After the statewide mandatory program begins it will no longer be necessary to use private laboratories for newborn screening. We solicit your cooperation and input as we develop our expansion plan.

Sincerely,

George Cunningham

Kathleen Velazquez

George Cunningham, M.D., M.P.H.
Chief, Genetic Disease Branch

Kathleen Velazquez, M.P.H., M.A.
Chief, Newborn Screening Section

Enclosures

Cc: Medical Directors and Project Directors,
Newborn Screening Area Service Centers

Medical Directors, CCS-Approved
Metabolic Centers

California Newborn Screening Program

Disorders Detectable by Tandem Mass Spectrometry (MS/MS)
Using Newborn Screening Dried Blood Spots (to be added to NBS Program mid-2005)

Primary Disorders (41)

Amino Acid Disorders (14)

- argininemia/arginase deficiency
- argininosuccinic acid lyase deficiency (ASAL deficiency)
- biopterin disorders (4)
- citrullinemia type I/argininosuccinic acid synthetase deficiency (ASAS deficiency)
- citrullinemia type II (citrin deficiency)
- homocystinuria/cystathionine beta-synthase deficiency (CBS deficiency)
- hypermethioninemia/MAT deficiency
- hyperphenylalaninemia – Classical Phenylketonuria (PKU)
- hyperphenylalaninemia – variant PKU
- hyperphenylalaninemia – benign PKU
- maple syrup urine disease – (MSUD)

Organic Acid Disorders (17)

- 2-methylbutyryl-CoA dehydrogenase deficiency
- 3-hydroxy-3-methylglutaryl-CoA lyase deficiency (HMGCoA lyase deficiency)
- 3-methylcrotonyl-CoA carboxylase deficiency (3MCC deficiency)
- 3-methylglutaconic aciduria (MGA) type I
- 3-methylglutaconic aciduria (MGA) type II
- 3-methylglutaconic aciduria (MGA) type III
- 3-methylglutaconic aciduria (MGA) type IV
- beta-ketothiolase deficiency (BKD)
- glutaric acidemia type-1 (GA-1)
- isobutyryl-CoA dehydrogenase deficiency
- isovaleric acidemia (IVA)
- methylmalonic acidemia , mut –
- methylmalonic acidemia, mut 0
- methylmalonic acidemia (Cbl A, B)
- methylmalonic acidemia (Cbl C, D)
- propionic acidemia (PA)
- multiple carboxylase deficiency (MCD)

Fatty Acid Oxidation Disorders (10)

- carnitine transporter deficiency
- carnitine-acylcarnitine translocase deficiency (CAT deficiency)
- carnitine palmitoyl transferase deficiency-type 1 (CPT-1 deficiency)
- carnitine palmitoyl transferase deficiency-type 2 (CPT-2 deficiency)
- long chain hydroxyacyl-CoA dehydrogenase deficiency (LCHAD deficiency)
- medium chain acyl-CoA dehydrogenase deficiency (MCAD deficiency)
- multiple acyl-CoA dehydrogenase deficiency (MAD deficiency)/glutaric acidemia type-2 (GA-2)
- short chain acyl-CoA dehydrogenase deficiency (SCAD deficiency)
- trifunctional protein deficiency (TFP deficiency)
- very long chain acyl-CoA dehydrogenase deficiency (VLCAD deficiency)

Secondary Disorders (13)

- 2-methyl-3-hydroxybutyryl-CoA dehydrogenase deficiency
- 5-oxoprolinuria

- ethylmalonic encephalopathy(EE)
- homocitrullinuria, hyperornithinemia, hyperammonemia -HHH
- gyrate atrophy of the choroid and retina
- malonic aciduria
- non-ketotic hyperglycinemia
- prolinemia type I
- prolinemia type II
- tyrosinemia type I (TYR-I)
- tyrosinemia type II (TYR-II)
- tyrosinemia type III (TYR-III)
- tyrosinemia, transient

California Department of Health Services
CCS-Approved Metabolic Centers and Medical Directors
(For PKU and other Metabolic Diseases)

Sutter Medical Center, Sacramento
5271 F Street, Bldg. C
Sacramento, CA 95819
(916) 733-6023 (Referral & Appointment)
Samuel Yang, M.D.

UC Davis Medical Center
2270 Stockton Boulevard
Sacramento, CA 95817
(916) 734-2011 (Main)
(916) 734-2107 (Appointment)
Dennis Styne, M.D.

UC San Francisco Medical Center
Third and Parnassus Avenue
San Francisco, CA 94143
(415) 476-1000 (Main)
(415) 476-2757 (Appointment)
Seymour Packman, M.D.

Children's Hospital & Research Center at Oakland
747 52nd Street
Oakland, CA 94609
(510) 428-3550 (Medical Genetics)
(510) 428-3885 ext 4401 (Dietician, Gail Seche)
John Waterson, M.D., Ph.D.

Kaiser Permanente Medical Center
280 West MacArthur Boulevard
Oakland, CA 94611
(510) 752-1000 (Main)
(510) 752-6298 (Genetic Dept.)
John Baker, M.D.

Lucile Salter Packard Children's Hospital at Stanford
770 Welch Road
Palo Alto, CA 94301
(650) 723-6858 (Main/Appointment)
Gregory Enns, M.B., Ch.B.

Children's Hospital of Central California
9300 Valley Children's Place
Madera, CA 93638
(559) 353-3000 (Main)
(559) 353-8800 (Appointment)
Susan Winter, M.D.

Cedars-Sinai Medical Center/Metabolic
444 S. San Vicente Blvd. #1001
Los Angeles, CA 90048
(310) 423-9993 (Main)
(310) 423-9914 (Appointment)
William Wilcox, M.D., Ph.D.

Children's Hospital of Los Angeles
4650 Sunset Blvd., #90
Los Angeles, CA 90027
(323) 660-2450 (Main)
(323) 669-2290 (Appointment)
Richard Boles, M.D.

Harbor-UCLA Medical Center
1000 West Carson St., Box 465
Torrance, CA 90509
(310) 222-3756 (Main/Appointment)
Adam Jonas, M.D.

Kaiser Southern California Metabolic Center
4700 Sunset Blvd, First Floor
Los Angeles, CA 90027
(323) 783-6970 (Main/Appointment)
Rebecca Mardach, M.D.

Los Angeles County-USC Medical Center
1129 North State St., Rm.1G-24
Los Angeles, CA 90033
(323) 226-3816 (Main/Appointment)
Atsuko Fujimoto, M.D., Ph.D.

UC Los Angeles Medical Center
10833 LeConte Avenue
Los Angeles, CA 90095-1752
(310) 206-6581 (Main)
(310) 825-0867 (Appointment)
Stephen D. Cederbaum, M.D.

UC Irvine Medical Center/Metabolic
101 City Drive South
Orange, CA 92868
(714) 456-7890 (Main)
(714) 456-6878 (Appointment)
James Bartley, M.D., Ph.D.

Children's Hospital, San Diego
3020 Children's Way
San Diego, CA 92123-4282
(619) 294-6104 (Metabolic Clinic)
(858) 966-5999 (Appointment)
Bruce Barshop, M.D.